

Column definitions

Exome

- X = index
- PPG_exoncount = processed pseudogene exon count
- SegDupProbeCount = segmental duplication probe count
- chromosome = chromosome
- familyID = Simons family id
- median_svdzrpkm = median normalized RPKM after SVD
- num_probes = number of probes in CNV
- rel = relationship of individual (f=father, m=mother, p=proband, s=sibling)
- sampleID = Simons sample id
- set = where CNV call originated (options are conifer, xhmm, or Intersection)
- size_bp = size in base pairs
- start = starting nucleotide
- state = state (1 is duplication, -1 is deletion)
- stdev_svdzrpkm = standard deviation of normalized RPKM after SVD
- stop = end nucleotide
- cnvr_frequency_SSC_merge = frequency of event in this dataset
- cnvrID_SSC_merge = cnvrID for this dataset
- inh = inheritance
- RefSeq = genes in the region

SNP microarray

- type = type of event (DUP=duplication, DEL=deletion)
- Sex = sex of individual
- Genotyping.Chip.Type = SNP microarray type that the sample was run on
- crlmm_number_snps = number of SNPs on microarray in CNV region
- crlmm_mean = mean copy number for the probes in the region from crlmm
- crlmm_percentileRankOfMean = percentile rank of the mean for the probes in the region from crlmm in comparison to the rest of the genome
- crlmm_pValue = p-value from permutation test to assess significance of the observed mean
- crlmm_1600_number_snps = number of SNPs on microarray in CNV region (border of 1,600 nucleotides)
- crlmm_1600_mean = mean copy number for the probes in the region from crlmm (border of 1,600 nucleotides)
- crlmm_1600_percentileRankOfMean = percentile rank of the mean for the probes in the region from crlmm in comparison to the rest of the genome (border of 1,600 nucleotides)
- crlmm_1600_pValue = p-value from permutation test to assess significance of the observed mean (border of 1,600 nucleotides)
- crlmm_2600_number_snps = number of SNPs on microarray in CNV region (border of 2,600 nucleotides)

- `crlmm _2600_mean` = mean copy number for the probes in the region from `crlmm` (border of 2,600 nucleotides)
- `crlmm _2600_percentileRankOfMean` = percentile rank of the mean for the probes in the region from `crlmm` in comparison to the rest of the genome (border of 2,600 nucleotides)
- `crlmm _2600_pValue` = p-value from permutation test to assess significance of the observed mean (border of 2,600 nucleotides)
- `crlmm _6000_number_snps` = number of SNPs on microarray in CNV region (border of 6,000 nucleotides)
- `crlmm _6000_mean` = mean copy number for the probes in the region from `crlmm` (border of 6,000 nucleotides)
- `crlmm _6000_percentileRankOfMean` = percentile rank of the mean for the probes in the region from `crlmm` in comparison to the rest of the genome (border of 6,000 nucleotides)
- `crlmm _6000_pValue` = p-value from permutation test to assess significance of the observed mean (border of 6,000 nucleotides)